glycogen storage disease type VI

Glycogen storage disease type VI (also known as GSDVI or Hers disease) is an inherited disorder caused by an inability to break down a complex sugar called glycogen in liver cells. A lack of glycogen breakdown interferes with the normal function of the liver.

The signs and symptoms of GSDVI typically begin in infancy to early childhood. The first sign is usually an enlarged liver (hepatomegaly). Affected individuals may also have low blood sugar (hypoglycemia) or a buildup of lactic acid in the body (lactic acidosis) during prolonged periods without food (fasting).

The signs and symptoms of GSDVI tend to improve with age; most adults with this condition do not have any related health problems.

Frequency

The exact prevalence of GSDVI is unknown. At least 11 cases have been reported in the medical literature, although this condition is likely to be underdiagnosed because it can be difficult to detect in children with mild symptoms or adults with no symptoms. GSDVI is more common in the Old Older Mennonite population, with an estimated incidence of 1 in 1,000 individuals.

Genetic Changes

Mutations in the *PYGL* gene cause GSDVI. The *PYGL* gene provides instructions for making an enzyme called liver glycogen phosphorylase. This enzyme is found only in liver cells, where it breaks down glycogen into a type of sugar called glucose-1-phosphate. Additional steps convert glucose-1-phosphate into glucose, a simple sugar that is the main energy source for most cells in the body.

PYGL gene mutations prevent liver glycogen phosphorylase from breaking down glycogen effectively. As a result, liver cells cannot use glycogen for energy. Since glycogen cannot be broken down, it accumulates within liver cells, causing these cells to become enlarged and dysfunctional.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- GSD type VI
- GSD VI
- GSD6
- hepatic glycogen phosphorylase deficiency
- Hers disease
- liver phosphorylase deficiency syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Glycogen storage disease, type VI
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0017925/

Other Diagnosis and Management Resources

- GeneReview: Glycogen Storage Disease Type VI
  https://www.ncbi.nlm.nih.gov/books/NBK5941

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Hepatomegaly
  https://medlineplus.gov/ency/article/003275.htm
- Health Topic: Carbohydrate Metabolism Disorders
  https://medlineplus.gov/carbohydratemetabolismdisorders.html

- Health Topic: Hypoglycemia
  https://medlineplus.gov/hypoglycemia.html
- Health Topic: Liver Diseases
  https://medlineplus.gov/liverdiseases.html

Genetic and Rare Diseases Information Center

- Glycogen storage disease type 6
  https://rarediseases.info.nih.gov/diseases/6529/glycogen-storage-disease-type-6

Educational Resources

- Cincinnati Children's Hospital: Glycogen Storage Disease
  https://www.cincinnatichildrens.org/health/g/gsd
- Disease InfoSearch: Glycogen Storage Disease Type 6
  http://www.diseaseinfosearch.org/Glycogen+Storage+Disease+Type+6/3126
- Merck Manual Consumer Version: Disorders of Carbohydrate Metabolism
  http://www.merckmanuals.com/home/children-s-health-issues/hereditary-metabolic-disorders/disorders-of-carbohydrate-metabolism
- Orphanet: Glycogen storage disease due to liver glycogen phosphorylase deficiency
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=369

Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases
  http://www.climb.org.uk/
- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/hers-disease/
- The Association for Glycogen Storage Disease (UK)
  http://www.agsd.org.uk/tabid/1138/default.aspx
- The Association for Glycogen Storage Disease (US)
  http://www.agsdus.org/type-vi.php
- University of Kansas Medical Center Resource List
  http://www.kumc.edu/gec/support/glycogen.html

GeneReviews

- Glycogen Storage Disease Type VI
  https://www.ncbi.nlm.nih.gov/books/NBK5941

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22glycogen+storage+disease+type+VI%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28glycogen+storage+disease+type+VI%5BALL%5D%29+OR+%28GSD+VI%5BALL%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

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